Docket No.: 2825.2012-004

Title: Association of Dopamine Beta-Hydra ese... inventors: Pamela Sklar, et al.

1: NM 000787. Homo sapiens dopam...[gi:4503260]

PubMed, Protein, Related Sequences, LinkOut

PRI 19-MAR-1999 NM 000787 2725 bp mRNA LOCUS

Homo sapiens dopamine beta-hydroxylase (dopamine DEFINITION

beta-monooxygenase) (DBH) mRNA.

NM 000787 ACCESSION

NM 000787.1 GI:4503260 VERSION

KEYWORDS

SOURCE human.

Homo sapiens ORGANISM

Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Mammalia;

Eutheria; Primates; Catarrhini; Hominidae; Homo.

(bases 1 to 2725) REFERENCE

Kobayashi, K., Kurosawa, Y., Fujita, K. and Nagatsu, T. **AUTHORS**

Human dopamine beta-hydroxylase gene: two mRNA types having TITLE

different 3'-terminal regions are produced through alternative

polyadenylation

Nucleic Acids Res. 17 (3), 1089-1102 (1989) **JOURNAL**

MEDLINE 89160241

(bases 1 to 2725) REFERENCE

Nagatsu, T. AUTHORS

Direct Submission TITLE

Submitted (14-OCT-1988) Nagatsu T., Department of Biochemistry, JOURNAL

Nagoya University, School of Medicine, Nagoya 466, Japan

REFSEQ: This reference sequence was derived from X13255. COMMENT

see also X13256 for type b mRNA

Map data from Craig et al. Cytogenet. Cell Genet. 48:48-50(1988).

PROVISIONAL RefSeq: This is a provisional reference sequence

record

that has not yet been subject to human review. The final curated reference sequence record may be somewhat different from this one.

Location/Oualifiers **FEATURES**

1..2725 source

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/db xref="taxon:9606"

/map="9q34"

1..2725 <u>qene</u>

/gene="DBH"

/db xref="LocusID: 1621"

/db xref="MIM:223360"

CDS 33..1844

/gene="DBH"

/EC number="1.14.17.1"

/codon start=1

/db xref="LocusID:1621"

/db xref="MIM:<u>223360</u>"

/product="dopamine beta-hydroxylase (dopamine

beta-monooxygenase) " /protein_id="NP_000778.1"

/db xref="GI:4503261"

/translation="MREAAFMYSTAVAIFLVILVAALQGSAPRESPLPYHIPLDPEGS

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SDQKGQIHLDPQQDYQLLQVQRTPEGLTLLFKRPFGTCDPKDYLIEDGTVHLVYGILE

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e: Association of Dopamine Beta-Hydroxyl entors: Pamela Sklar, et al.

EPFRSLEAINGSGLQMGLQRVQLLKPNIPEPELPSDACTMEVQAPNIQIPSQETTYWC
YIKELPKGFSRHHIIKYEPIVTKGNEALVHHMEVFQCAPEMDSVPHFSGPCDSKMKPD
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IRLYYTAKLRRFNAGIMELGLVYTPVMAIPPRETAFILTGYCTDKCTQLALPPSGIHI
FASQLHTHLTGRKVVTVLVRDGREWEIVNQDNHYSPHFQEIRMLKKVVSVHPGDVLIT
SCTYNTEDRELATVGGFGILEEMCVNYVHYYPQTQLELCKTAVDAGFLQKYFHLINRF
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siq peptide 33..107
mat peptide 108..1841

/product="dopamine beta-hydroxylase (dopamine

beta-monooxygenase)"

517 t 533 a 774 q 901 c BASE COUNT ORIGIN 1 tcagtcgctg ggccagcctg cccggcccca gcatgcggga ggcagccttc atgtacagca 61 cagcagtggc catcttcctg gtcatcctgg tggccgcact gcagggctcg gctccccgtg 121 agagecect ecectateae ateceeetgg acceggaggg gteeetggag eteteatgga 181 atgtcagcta cacccaggag gccatccatt tccagctcct ggtgcggagg ctcaaggctg 241 gcgtcctgtt tgggatgtcc gaccgtggcg agcttgagaa cgcagatctc gtggtgctct 301 ggaccgatgg ggacactgcc tattttgcgg acgcctggag tgaccagaag gggcagatcc 361 acctggatcc ccagcaggac taccagctgc tgcaggtgca gaggacccca gaaggcctga 421 ccctgctttt caagaggccc tttggcacct gcgaccccaa ggattacctc attgaagacg 481 gcactgtcca cttggtctac gggatcctgg aggagccgtt ccggtcactg gaggccatca 541 acggctcggg cctgcagatg gggctgcaga gggtgcagct cctgaagccc aatatccccg 601 aaccggagtt gccctcagac gcgtgcacca tggaggtcca agctcccaat atccagatcc 661 ccagccagga gaccacgtac tggtgctaca ttaaggagct tccaaagggc ttctctcggc 721 accacattat caagtacgag cccatcgtca ccaagggcaa tgaggccctt gtccaccaca 781 tggaagtett ccagtgegee eccgagatgg acagegtee ccaetteage gggeeetgeg 841 actccaagat gaaacccgac cgcctcaact actgccgcca cgtgctggcc gcctgggccc 901 tgggtgccaa ggcattttac tacccagagg aagccggcct tgccttcggg ggtccagggt 961 cctccagata tctccgcctg gaagttcact accacaaccc actggtgata gaaggacgaa 1021 acgactecte aggeatecge ttgtactaca cagecaaget geggegette aacgegggga 1081 tcatggagct gggactggtg tacacgccag tgatggccat tccaccacgg gagaccgcct 1141 tcatcctcac tggctactgc acggacaagt gcacccagct ggcactgcct ccctccggga 1201 tocacatott ogcototoag otocacacac acotgactgg gagaaaggtg gtcacagtgc 1261 tggtccggga cggccgggag tgggagatcg tgaaccagga caatcactac agccctcact 1321 tocaggagat cogcatgttg aagaaggtog tgtoggtoca tocgggagat gtgotoatoa 1381 cctcctgcac gtacaacacg gaagaccggg agctggccac agtggggggc ttcgggatcc 1441 tggaggagat gtgtgtcaac tacgtgcact actaccccca gacgcagctg gagctctgca 1501 agacggctgt ggacgccggc ttcctgcaga agtacttcca cctcatcaac aggttcaaca 1561 acgaggatgt ctgcacctgc cctcaggcgt ccgtgtctca gcagttcacc tctgttccct 1621 ggaactcctt caaccgcgac gtactgaagg ccctgtacag cttcgcgccc atctccatgc 1681 actgcaacaa gtcctcagcc gtccgcttcc agggtgaatg gaacctgcag cccctgccca 1741 aggtcatctc cacactggaa gagcccaccc cacagtgccc caccagccag ggccgaagcc

Figure 1B

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le: Association of Dopamine Beta-Hydroxy ventors: Pamela Sklar, et al.

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1801 ctgctggccc caccgttgtc agcattggtg ggggcaaagg ctgagggggg acctactcct
1861 ccccctcctc catgctgtcc ctgtgggctc acaccggcac tgtgcactct actctgcgac
1921 gatececatg gaacageest geacgeecag gatgaagggg ceagaceaeg eeeetgeetg
1981 agaccacggt ccaatccagc cttcttcccc cagggtcccc tgcatggctg agagggtgtg
2041 ggtgccctgt tgacctaccc tggaccgagt ggaccacgac ctcgtccatt taaacccggc
2101 tgactcagtg cagggacagc ccgcacagtg gtccagggtc cagccctccg ccagccctgt
2161 tecgeeteae tgggtgtgge etggettetg ggacaggeae catgetggge eggggtgtgg
2221 aatcaccggg aacgcccccg cccccgcccc gctgctcccg gtgtgcagcg ggtgcgggtg
2281 ccgcttaaac atttccctgc tgagtggctc gtgtttcaca gtgggcggct tccctgcgac
2341 ggaggcagga ccaggcattt agctagttag agactcgcct gggaaattgc tccattcctg
2401 agtaaacaga tattttcgcc cacctaaagg gaagccctga caacaactat caccaaaaga
2461 cgaggcggca aagatccagc ggggcttctg ggcgccggtt ccacgtgggg tggaattatt
2521 agcaccaget tgcttctctg ccggtggggc cagcgctgaa cagaccgggg tggagtcagg
2581 gctgtgcttt ccgcgtggtt ctgccactta gggagtgtgc cttgggcggg ccatttcaca
2641 ttcctgaccc tcacttttct catctgtaaa accaggctga tgccgtgcgg gctaatgagc
2701 caataaagct cacacttggg ctggc
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Figure 1C

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Title: Associa	tion of Dopamine Beta-Hydr
Inventors:	Pamela Sklar, et al.

	SUMMARY OF DBH ASSOCIATION	MATION		
Original study	Transmilled	Transmilled Uniransmitted	Chl-sq	p-val
DBHu2	20	2.1	0.63	0.4658
(DBHu1	18	12	1.20	0.2733
DBHp444a	56	4	2.32	0.1278
Replication/DePaulo				
DBHu2	14	€	1.64	0.2008
DBHn1		13	0.17	0.6831
DBHp444a	949	3.8	1.39	0,2383
Totals				
DBHu2	40	53	1.75	0.1854
DBHut	28	26	0.30	0.5882
DBHp444a	105	7.9	3.67	0.0653

		22.5		
Orginal Study	Transmitted	Transmitted Untransmitted	Chi-sq	p-yal
Allelet from iDBHu2 and ut	37	22	3.81	0.0508
Allele 1 from DBHu2 and DBHp444a	4 6	6.04	3.40	0.0629
Allele1 from DBHu1 and DBHp444a	₽	23	6.08	0.0138
Alielat from all three SNPs	46	25	6.21	0.0127
Replication/DePaulo				
Allelet from DBHu2 and ut	18	-1	1.69	0.1936
Aliele1 from DBHu2 and DBHp4446	32	14	7.04	0.0080
Allele1 from DBHu1 and DBHp444a	31	16	4.79	0.0287
Allele 1 from all three SNPs	31	11	9.52	0.0020
Totals				
Alleie1 from DBHu2 and u1	55	33	5.50	0.0180
Allele1 from DBHu2 and DBHp4448	11	43	9.63	0.0019
Allele1 from DBHu1 and DBHp444a	7.4	38	10.84	0.0010
Allele 1 from all three SNPs	77	90	14.88	0.0001